

## Comparative Analysis of Direct to Consumer Genetic Testing Kits

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### Abstract

Direct to consumer (DTC) genetic testing kits are becoming increasingly popular; as a result concerns about what results mean for consumers are showing up in the media. The purpose of this study was to evaluate DTC genetic testing kits to determine inter-test reliability. The companies of interest included Ancestry.com, 23andMe, and MyHeritage. 16 participants, 9 females and 7 males representing a variety of ethnic groups took the three independent DTC tests for ancestry lineage. Collection procedures specified by each company were used. Results for each participant were compared across companies, with similarities and discrepancies in identical categories being noted. All of the participants had major discrepancies within the data sets. For example, 23andMe reported 93.7% North and Western European ancestry for one participant while MyHeritage reported 25.0% North and Western European. Ancestry.com reported 53.0% East African ancestry while 23andMe reported 85.9%. Ancestry.com reported 42.0% Iberian ancestry for a participant while 23andMe reported 76.5%; for another participant Ancestry.com reported 9.0% Nigerian ancestry while MyHeritage reported 46.5%; and 23andMe reported 7.8% Japanese ancestry for a participant while MyHeritage reported 49.4%. These discrepancies point out the unreliability of this consumer science and can create complications for individuals who assume that the one DTC test they used is an accurate portrayal of their ancestry, and therefore their identity in one or more ethnic groups. Work needs to be done to ensure that consumer DNA reports are indeed accurate reflections of genetic ancestry.

**Keywords:** Ancestry, Direct to Consumer, AIMs

### 1. Introduction

Direct to consumer (DTC) genetic testing kits originated in 2000 by Family Tree DNA, and by the year 2020, these kits had grown into a multi-billion dollar industry with close to 40 individual DTC genetic testing kits available on the market. There are many reasons why an individual might want to take a DTC genetic ancestry test, ranging from curiosity to a desire to learn about their personal history and culture. Most individuals only purchase one kit and assume that the results obtained are accurate and portray their ancestry appropriately. In some cases, individuals use this information to change the perceptions they have of self and family, without knowing the accuracy of the results.

An investigative report performed by *The Doctors* in 2017, which used three sets of identical triplets to test various ancestry DNA kits, found that two out of the three sets of triplets had at least some results that differed from their siblings even though they share the same genetic makeup<sup>1</sup>. This study demonstrates that even with the large advancements made in genetic testing, all results cannot be taken at face value.

The process in which all DTC genetic testing companies compare customer samples is through their reference databases known as Ancestry Informative Markers (AIMs)<sup>2</sup>. Essentially, the more AIMs that are used for estimates of genome ancestry the more accurate the estimate is presumed to be<sup>3</sup>. AIMs produce the percentages of ancestry for each individual by comparing the DNA collected to volunteers who have provided DNA samples to these companies since their first days in business. Since the samples come from volunteers, and each company has amassed its collection of volunteers, the pool of genetic information that is being used to conclude ancestry is different for each company. Add to this the fact that the computer algorithms used to project backward are carefully guarded secrets and

developed independently by each company and it becomes clear that there may be a lot of subjectivity in the answers provided to consumers.

Jennifer Wagner<sup>4</sup> described some hypothetical scenarios where major issues may arise from consumers performing the DTC genetic ancestry testing kits. One such scenario explores the cultural implications of an African American man who, growing up, did not know his paternal lineage. After taking an ancestry testing kit he learns that he has some European ancestry on his paternal side of the family. In this scenario, the outcome wasn't too bad, but Wagner notes that given the large numbers of individuals looking at results from their tests some might become very unhappy about what they see. For instance, what happens when the DNA report of ancestry differs from the oral tradition within a family? Which is more reliable? DNA (even if we don't know how the answers were calculated) or first-hand family knowledge?

The purpose of this study was to compare three of the most popular DTC genetic testing kits, using the same set of volunteers, to determine what similarities and differences exist in the reports produced. Based on the study of triplets published in 2010<sup>1</sup> and a preliminary study reported from this lab<sup>5</sup>, a hypothesis was established that most individuals tested would have major differences in at least some ethnic backgrounds and that these differences would be seen regardless of the overall ethnicity of the individuals.

## 2. Materials & Methods

A total of 16 individuals representing a variety of ethnic backgrounds were recruited to take the DTC genetic tests. With the approval of the university IRB, each individual signed a release form agreeing to share the complete ancestry reports with the investigators. Reports were kept confidential from individuals outside the research group.

Test kits were purchased from Ancestry.com, MyHeritage, and 23andMe. Each individual in the study was given a set of three test kits when they signed the release forms. Individuals were instructed in the use of the kits; sampling and shipping procedures varied by company. Each individual obtained his/her samples according to the published procedures and mailed the kits back to the labs. Reports were generally available electronically and most individuals chose to forward those results to the investigators electronically, where they were printed out and filed. A few individuals brought paper copies of the reports to the lead investigator. Once all three reports were obtained for an individual the reports were gone over and similarities and differences between them were noted. Data points were chosen by cross-referencing each participant's percentage of ancestry within a single ethnic or geographic group from all three companies. Discrepancies were based on the labeling that each DTC genetic testing company used for their regions; if labeling did not match completely then general geographical locations were used to match percentages together. Data points that had a difference of more than 5% between the three reports were considered a discrepancy. This metric is the general standard for any statistical deviation within the natural sciences. Deviation in this study is defined by the difference between the percentages of an ethnic ancestry reported by each of the DTC genetic ancestry testing companies.

## 3. Results

Table 1 presents a percentage of an ethnic group that applies to the individual participant across the three direct to consumer genetic testing companies which are 23andMe, Ancestry.com, and MyHeritage. This table includes 16 participants with a variety of self-reported ethnicities. Table 2 illustrates the same percentages as in Table 1 but also includes the standard deviation for each company along with the mean for each company's data set. The standard deviation for 23andMe is 26.7, Ancestry.com has a standard deviation of 28.3, and MyHeritage has a standard deviation of 20.3. The mean for 23andMe is 66.6, Ancestry.com has a mean of 52.5, and MyHeritage has a mean of 59.3. Table 3 demonstrates 9 T-Tests to discover if there is a significant difference between each variable. The Ancestry.com-23andMe value is 0.156, the MyHeritage-23andMe value is 0.386, and the MyHeritage-Ancestry.com value is 0.444. These values are statistically insignificant because the T-Test values are above 0.05. Figure 1 shows the average differences between each DTC genetic testing company in graph form. It displays the mean on the Y-axis and the three companies along the X-axis. The graph also has standard error bars. Figure 2 shows how the three companies reflect consumer's results via a geographic map. The one on the left side is from the company MyHeritage, the top right is from the company 23andMe, and the bottom right is from the company Ancestry.com. These images are indicative of a single individual which is participant 2 in Table 1. Figure 3 is an image taken from the DNA results

summary of Ancestry.com where when the ethnicity estimate is selected, a range appears for that ethnicity. The range in Figure 3 is from 0-36% and the range amount varies by individual and ethnicity.

Table 1: List of 16 major discrepancies for each participant between each DTC genetic ancestry testing company along with their self-identified ethnicity

Participant	Sex	Self-Reported Ethnicity	23andMe	Ancestry.com	MyHeritage
1	F	Hispanic	76.5% Iberian	42.0% Iberian	59.6% Iberian
2	F	Asian	7.8% Japanese	100% East Asia	49.4% Japanese
3	F	Caucasian	93.7% North and West European	25.0% North and West European	88.5% North and West European
4	M	Caucasian	35.6% British and Irish	51.0% England Wales and Northwestern Europe	23.3% English
5	F	Caucasian	36.5% British and Irish	59% England Wales and Northwestern Europe	21.6% English, Scottish, English, & Wales
6	F	West African/ Guinean	47.4% Coastal West African	92.0% Mali	56.9% West African
7	M	East African/ Somali	99.5% Somali	38.0% Africa South Central Hunter Gathers	91.4% Somali
8	M	West African/ Senegalese	88.1% Guinean	50.0% Mali	71.4% West African
9	M	West African/ Guinean	68.0% Guinean	79.0% Mali	54.6% West African
10	F	East African/ Tanzanian & Kenyan	85.9% East African	53.0% East African	70.0% Kenyan
11	M	Central African/ Cameroon	37.3% Congolese	100.0% Cameroon	69.1% Nigerian
12	F	African American	67.8% West African	13.0% Nigerian	46.6% Nigerian
13	M	African American	81.8% West African	9.0% Nigerian	46.5% Nigerian
14	F	African American	62.0% Sub-saharan African	30.0% Cameroon, Congo and Southern Bantu	45.8% West African
15	F	Caucasian	82.2% North and West European	41.0% North and West European	76.7% North and West European
16	M	Caucasian	96.0% North and West European	58.0% North and West European	76.7% North and West European

Table 2: T-test Values between the different DTC genetic testing companies.

T-Test	23andMe	Ancestry.com	MyHeritage
23andMe	X	0.156	0.386
Ancestry.com	0.156	X	0.444
MyHeritage	0.386	0.444	X

Table 3: Standard deviation and Means for all participants and their ancestry reports.

	23andMe	Ancestry.com	MyHeritage
Participant 1	76.5	42	59.6
Participant 2	7.8	100	49.4
Participant 3	93.7	25	88.5
Participant 4	35.6	51	23.3
Participant 5	36.5	59	21.6
Participant 6	47.4	92	56.9
Participant 7	99.5	38	91.4
Participant 8	88.1	50	71.4
Participant 9	68	79	54.6
Participant 10	85.9	53	70
Participant 11	37.3	100	69.1
Participant 12	67.8	13	46.6
Participant 13	81.8	9	46.5
Participant 14	62	30	45.8
Participant 15	82.2	41	76.7
Participant 16	96	58	76.7
Mean	66.6	52.5	59.3
Standard Deviation	26.7	28.3	20.3

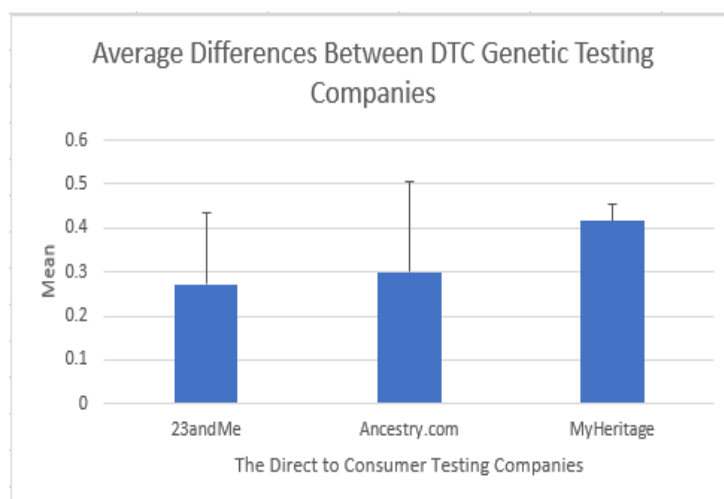


Figure 1: graph comparison of the means between the three DTC genetic testing kits.

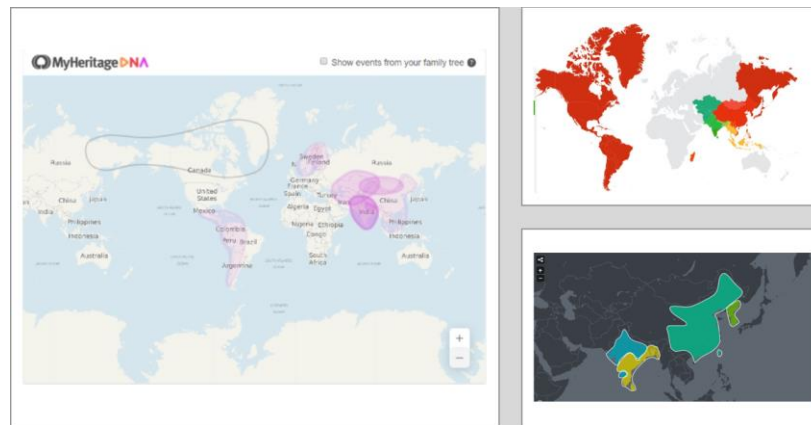


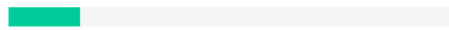
Figure 2: Geographic map of participant 2 from MyHeritage (left), 23andMe (top right), and Ancestry.com (bottom right).

### Germanic Europe

Primarily located in: Germany, Switzerland

[Learn more about this map and ethnicity](#)

16%



Your ethnicity estimate is 16%, but it can range from 0–36%. ⓘ

Figure 3: Range is shown for one participant Germanic European Ancestry.com report.

## 4. Discussion

DTC genetic ancestry testing kits have only been available to the public for about twenty years. During this time each company has gathered its reference samples and produced its computer algorithms for determining ethnic and/or geographic ancestry. Various assumptions have been made by scientists and computer programmers; since the reference DNA comes from living people and not ancient ancestors, these assumptions are very important and are derived from the available pool of evidence, which changes over time.

DTC genetic ancestry kits are not inexpensive (ranging from \$49. to over \$500.). Although no data can be found about consumer purchases, it is assumed that the vast majority of consumers purchase (and therefore rely on the reporting from) a single test. If all of the tests were equivalent that would not be a problem, but if there are differences in assumptions made within the algorithms used by each company then that can become an issue. In the current study, it was found that discrepancies occurred between reports received by all 16 participants, thus making this a very real concern. Even if one looks at all three reports together it is often hard to get a true picture of ancestry.

This poses uncertainty for the regular consumer because of how each company reports its findings. Each company has some similarities when it comes to reporting large swaths of geographic area, for example, North and West Europe, East Asia, and the Middle East. But when each company tries to report ethnicities into more specific areas such as Germanic Europe, specific African countries, and Native American tribes, the DTC genetic ancestry companies become wildly variable. Due to these inconsistencies, this made data analysis very difficult. In addition to the inconsistent mapping of gene pools, the error range within the companies' own reports was staggering. 23andMe, along with MyHeritage, do not even report a range of ancestry for the consumer to use in further analysis of the information presented to them. Ancestry.com did report a range that was as small as 1-5%-points to as large as 36%-

point range for an individual's ancestry as seen in Figure 2. This suggests all three companies have ranges concerning the number reported to the consumer.

Because these tests are advertised as scientifically-based and accurate (no error rates reported by any company), it is safe to assume that most consumers will accept whatever the report shows even if it does not match what has been handed down through oral tradition within the family. This is evidenced by Ancestry.com's own television commercial, in which a gentleman who thought he was Scottish (from family tradition) found out he was German (from the test results) and "changed his kilt for lederhosen". In other cases, the results might cause confusion or even in-fighting within a family. In other words, how individuals or even families see themselves ethnically may indeed depend on which particular ancestry DNA test is used.

As individuals fill out Census Bureau forms, college applications, and many other federal or state forms, deciding which ethnicity box to check may depend not only on family heritage but, increasingly, on reports received from various DTC genetic testing companies. Since these reports can lead individuals to new and sometimes very different conclusions about themselves and their families, it is important that consumers know and understand the limitations of these tests and the still-growing science of genetic ancestry testing. It is also important that these companies share data so that the best, scientifically-accurate, information is available to all.

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